Human embryonic stem cells give clues to Huntington's disease origins

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Researchers in Australia studying human embryonic stem cells have found evidence of the always-lethal Huntington's disease when the cells are just a few days old. The disease, caused by a mutation in a single gene, normally starts causing symptoms when people are in mid-life.

This research shows the value of being able to study how a disease progresses in a lab dish. It would normally not be possible extract brain cells from people and study them in a lab dish. But in this case the scientists started with IVF embryos that contained the Huntington's disease mutation, given by people using IVF to screen for the mutation before implantation. The embryos would otherwise be destroyed. They could extract embryonic stem cells, and then planned to mature those cells into the types of neurons damaged in Huntington's disease as a way of understanding how the disease forms.

What the group learned is that the first indication of a problem is visible within days, in the mitochondria that generate power for the cell. An ABC story quotes senior author Leon McQuade from the Macquarie University's Australian Proteome Analysis Facility (APAF) in Sydney as saying:

> "Human embryonic stem cells provide us with a very good model for doing drug toxicology and efficacy testing - it's a model that we really haven't had before."

The scientists can use these cells to screen for drugs that might one day treat the disease.

CIRM funds two research projects, both being carried out by University of California, Davis scientists, working toward new therapies for Huntington's Disease (links to research projects available here). One is generating new stem cell lines that contain the disease-causing mutation, such as those used in the Australian study. The other is working toward a therapy involving implanting a form of tissue specific stem cell into people with Huntington's disease.

Learn more about CIRM work towards a stem cell based therapy for Huntington's Disease on our Huntington's Disease Stem Cell Fact Sheet.

Here's more on Huntington's disease and the search for a cure:

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